Penelope E Bonnen

List of Publications by Year
in descending order

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Version: 2024-02-01

47 papers	6,354 citations	186265 28 h-index	214800 47 g-index
48 all docs	48 docs citations	48 times ranked	14219 citing authors

#	Article	IF	Citations
1	Integrating common and rare genetic variation in diverse human populations. Nature, 2010, 467, 52-58.	27.8	2,625
2	Autosomal dominant cerebellar ataxia (SCA6) associated with small polyglutamine expansions in the Î±1A-voltage-dependent calcium channel. Nature Genetics, 1997, 15, 62-69.	21.4	1,606
3	Translational control of mGluR-dependent long-term depression and object-place learning by eIF2Î±. Nature Neuroscience, 2014, 17, 1073-1082.	14.8	159
4	Mutations in FBXL4 Cause Mitochondrial Encephalopathy and a Disorder of Mitochondrial DNA Maintenance. American Journal of Human Genetics, 2013, 93, 471-481.	6.2	137
5	Identification of Variant-Specific Functions of PIK3CA by Rapid Phenotyping of Rare Mutations. Cancer Research, 2015, 75, 5341-5354.	0.9	130
6	The GABA Transaminase, ABAT, Is Essential for Mitochondrial Nucleoside Metabolism. Cell Metabolism, 2015, 21, 417-427.	16.2	119
7	Recurrent Muscle Weakness with Rhabdomyolysis, Metabolic Crises, and Cardiac Arrhythmia Due to Bi-allelic TANGO2 Mutations. American Journal of Human Genetics, 2016, 98, 347-357.	6.2	98
8	Recurrent De Novo Dominant Mutations in SLC25A4 Cause Severe Early-Onset Mitochondrial Disease and Loss of Mitochondrial DNA Copy Number. American Journal of Human Genetics, 2016, 99, 860-876.	6.2	93
9	LRPPRCmutations cause early-onset multisystem mitochondrial disease outside of the French-Canadian population. Brain, 2015, 138, 3503-3519.	7.6	81
10	Apparent underdiagnosis of Cerebrotendinous Xanthomatosis revealed by analysis of ~60,000 human exomes. Molecular Genetics and Metabolism, 2015, 116, 298-304.	1.1	79
11	Haplotype and Linkage Disequilibrium Architecture for Human Cancer-Associated Genes. Genome Research, 2002, 12, 1846-1853.	5.5	78
12	Mitochondrial Disease Sequence Data Resource (MSeqDR): A global grass-roots consortium to facilitate deposition, curation, annotation, and integrated analysis of genomic data for the mitochondrial disease clinical and research communities. Molecular Genetics and Metabolism, 2015, 114, 388-396.	1.1	76
13	First Complete Genome Sequence of Two Staphylococcus epidermidis Bacteriophages. Journal of Bacteriology, 2007, 189, 2086-2100.	2.2	71
14	Mutations in MDH2, Encoding a Krebs Cycle Enzyme, Cause Early-Onset Severe Encephalopathy. American Journal of Human Genetics, 2017, 100, 151-159.	6.2	63
15	Evaluating potential for whole-genome studies in Kosrae, an isolated population in Micronesia. Nature Genetics, 2006, 38, 214-217.	21.4	61
16	Haplotypes at ATM Identify Coding-Sequence Variation and Indicate a Region of Extensive Linkage Disequilibrium. American Journal of Human Genetics, 2000, 67, 1437-1451.	6.2	56
17	De Novo Mutations in SLC25A24 Cause a Craniosynostosis Syndrome with Hypertrichosis, Progeroid Appearance, and Mitochondrial Dysfunction. American Journal of Human Genetics, 2017, 101, 833-843.	6.2	56
18	<scp>OXA</scp> 1L mutations cause mitochondrial encephalopathy and a combined oxidative phosphorylation defect. EMBO Molecular Medicine, 2018, 10, .	6.9	54

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19	SCYL1 variants cause a syndrome with lowÎ³-glutamyl-transferase cholestasis, acute liver failure, and neurodegeneration (CALFAN). Genetics in Medicine, 2018, 20, 1255-1265.	2.4	50
20	Phenotype of GABA-transaminase deficiency. Neurology, 2017, 88, 1919-1924.	1.1	49
21	Clinical, morphological, biochemical, imaging and outcome parameters in 21 individuals with mitochondrial maintenance defect related to FBXL4 mutations. Journal of Inherited Metabolic Disease, 2015, 38, 905-914.	3.6	45
22	TM4SF20 Ancestral Deletion and Susceptibility to a Pediatric Disorder of Early Language Delay and Cerebral White Matter Hyperintensities. American Journal of Human Genetics, 2013, 93, 197-210.	6.2	43
23	Inhibition of Upf2-Dependent Nonsense-Mediated Decay Leads to Behavioral and Neurophysiological Abnormalities by Activating the Immune Response. Neuron, 2019, 104, 665-679.e8.	8.1	43
24	WDR35 mutation in siblings with Sensenbrenner syndrome: A ciliopathy with variable phenotype. American Journal of Medical Genetics, Part A, 2012, 158A, 2917-2924.	1.2	40
25	Bi-allelic ADPRHL2 Mutations Cause Neurodegeneration with Developmental Delay, Ataxia, and Axonal Neuropathy. American Journal of Human Genetics, 2018, 103, 817-825.	6.2	40
26	Pathogenic Bi-allelic Mutations in NDUFAF8 Cause Leigh Syndrome with an Isolated Complex I Deficiency. American Journal of Human Genetics, 2020, 106, 92-101.	6.2	39
27	Clinical, biochemical, and genetic features of four patients with shortâ€chain enoylâ€CoA hydratase (ECHS1) deficiency. American Journal of Medical Genetics, Part A, 2018, 176, 1115-1127.	1.2	36
28	Successful diagnosis of HIBCH deficiency from exome sequencing and positive retrospective analysis of newborn screening cards in two siblings presenting with Leigh's disease. Molecular Genetics and Metabolism, 2015, 115, 161-167.	1.1	32
29	Mutations inELAC2associated with hypertrophic cardiomyopathy impair mitochondrial tRNA 3â€²â€end processing. Human Mutation, 2019, 40, 1731-1748.	2.5	31
30	Bi-allelic HPDL Variants Cause a Neurodegenerative Disease Ranging from Neonatal Encephalopathy to Adolescent-Onset Spastic Paraplegia. American Journal of Human Genetics, 2020, 107, 364-373.	6.2	30
31	Pathogenic variants in HTRA2 cause an earlyâ€onset mitochondrial syndrome associated with 3â€methylglutaconic aciduria. Journal of Inherited Metabolic Disease, 2017, 40, 121-130.	3.6	23
32	Clinical, biochemical, and genetic features associated with VARS2 -related mitochondrial disease. Human Mutation, 2018, 39, 563-578.	2.5	22
33	Loss-of-function mutations inISCA2disrupt 4Fe-4S cluster machinery and cause a fatal leukodystrophy with hyperglycinemia and mtDNA depletion. Human Mutation, 2018, 39, 537-549.	2.5	21
34	FBXL4-Related Mitochondrial DNA Depletion Syndrome 13 (MTDPS13): A Case Report With a Comprehensive Mutation Review. Frontiers in Genetics, 2019, 10, 39.	2.3	21
35	POLRMT mutations impair mitochondrial transcription causing neurological disease. Nature Communications, 2021, 12, 1135.	12.8	21
36	Incidence of PKAN determined by bioinformatic and population-based analysis of ~140,000 humans. Molecular Genetics and Metabolism, 2019, 128, 463-469.	1.1	20

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37	LONP1 de novo dominant mutation causes mitochondrial encephalopathy with loss of LONP1 chaperone activity and excessive LONP1 proteolytic activity. Mitochondrion, 2020, 51, 68-78.	3.4	16
38	From incomplete penetrance with normal telomere length to severe disease and telomere shortening in a family with monoallelic and biallelic PARN pathogenic variants. Human Mutation, 2019, 40, 2414-2429.	2.5	14
39	POLG2 deficiency causes adultâ€onset syndromic sensory neuropathy, ataxia and parkinsonism. Annals of Clinical and Translational Neurology, 2017, 4, 4-14.	3.7	13
40	Novel compound mutations in the mitochondrial translation elongation factor (TSFM) gene cause severe cardiomyopathy with myocardial fibro-adipose replacement. Scientific Reports, 2019, 9, 5108.	3.3	12
41	European admixture on the Micronesian island of Kosrae: lessons from complete genetic information. European Journal of Human Genetics, 2010, 18, 309-316.	2.8	11
42	Cerebrotendinous Xanthomatosis Presenting with Infantile Spasms and Intellectual Disability. JIMD Reports, 2016, 35, 1-5.	1.5	10
43	Human COQ4 deficiency: delineating the clinical, metabolic and neuroimaging phenotypes. Journal of Medical Genetics, 2022, 59, 878-887.	3.2	9
44	<scp>TAB2</scp> variants cause cardiovascular heart disease, connective tissue disorder, and developmental delay. Clinical Genetics, 2022, 101, 214-220.	2.0	7
45	RRM1 variants cause a mitochondrial DNA maintenance disorder via impaired de novo nucleotide synthesis. Journal of Clinical Investigation, 2022, 132, .	8.2	6
46	Metabolomics Profile in ABAT Deficiency Pre- and Post-treatment. JIMD Reports, 2018, 43, 13-17.	1.5	5
47	Longitudinal study shows increasing obesity and hyperglycemia in micronesia. Obesity, 2013, 21, E421-7.	3.0	3